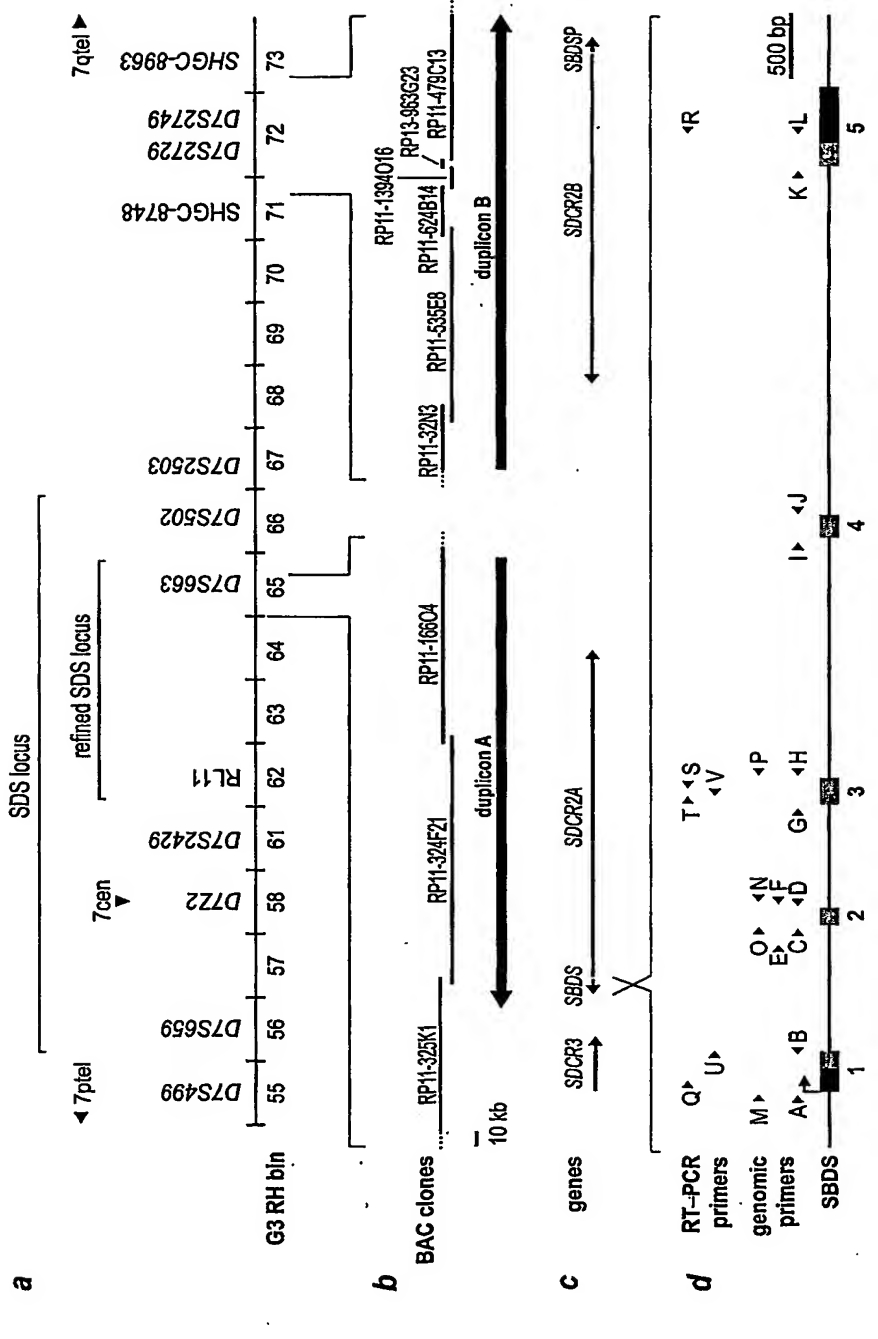

a
b
c
d
SDS locus
refined SDS locus
7ptel
7cen
7qtel
SHGC-8963
D7S2749
D7S2729
SHGC-8748
D7S2503
D7S502
D7S663
RL11
D7S2429
D7Z2
D7S659
D7S499
G3 RH bin
RP11-325K1
RP11-324F21
RP11-166O4
RP11-139A016
RP13-963G23
RP11-479C13
RP11-624B14
RP11-535E8
RP11-32N3
duplicon A
duplicon B
10 kb
BAC clones
SBDS
SDCR3
SDCR2A
SDCR2B
SBDSP
genes
RT-PCR primers
genomic primers
SBDS
500 bp

FIGURE 1

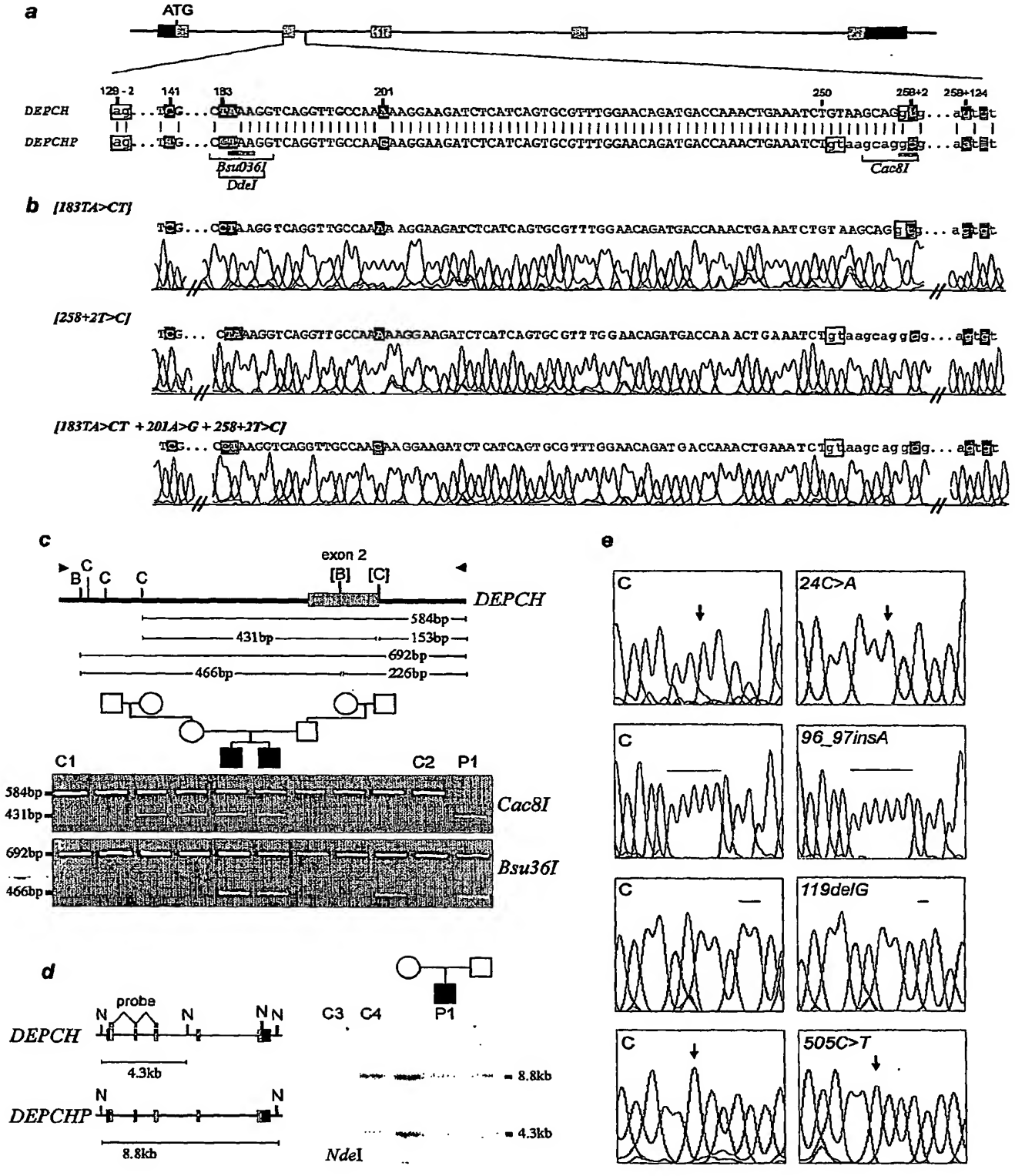

a
ATG
DEPCH
DEPCHP
Bsu036I
DdeI
Cac8I
b
[183TA>CT]
[258+2T>C]
[183TA>CT + 201A>G + 258+2T>C]
c
exon 2
DEPCH
584bp
431bp
153bp
692bp
466bp
226bp
C1
C2
P1
Cac8I
Bsu36I
d
probe
DEPCH
4.3kb
DEPCHP
8.8kb
C3
C4
P1
8.8kb
4.3kb
NdeI
e
C
24C>A
96_97insA
119delG
505C>T

FIGURE 2

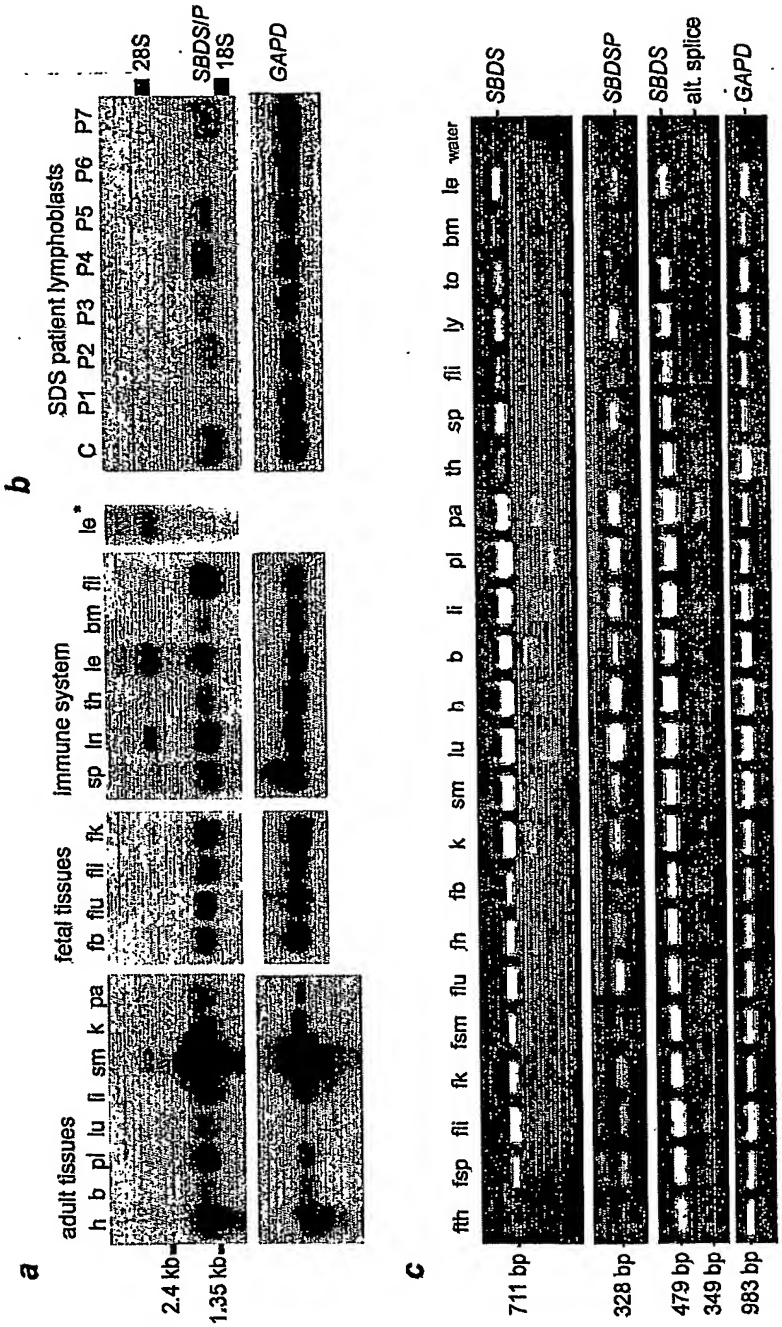

a
adult tissues
h b pl lu li sm k pa
fetal tissues
fb flu fil fk
immune system
sp ln th le bm fli
le*
2.4 kb
1.35 kb
b
SDS patient lymphoblasts
C P1 P2 P3 P4 P5 P6 P7
28S
18S
SBDSIP
GAPD
c
fth fsp fli fk fsm flu fh fb k sm lu h b li pl pa th sp fli ly to bm le water
SBDS
SBDSP
SBDS alt. splice
GAPD
711 bp
328 bp
479 bp
349 bp
983 bp

FIGURE 3


N8K
N34fs15
S41fs15
E44G
K62X
K67E
84Cfs3
I87S
D97_K98delinsEVQVS
R126T
R169C
I126T
U1-like zinc finger
Ath
Dme
Cel
Mmu
Hsa
Ola
Sce
Ecu
Mac
Hnr
Mth
Mka
Mja
Afu
Pab
Tac
Pae
Sso
Ape
Gar+
Pba+

FIGURE 4

SBDS cDNA Sequence ID NO:1

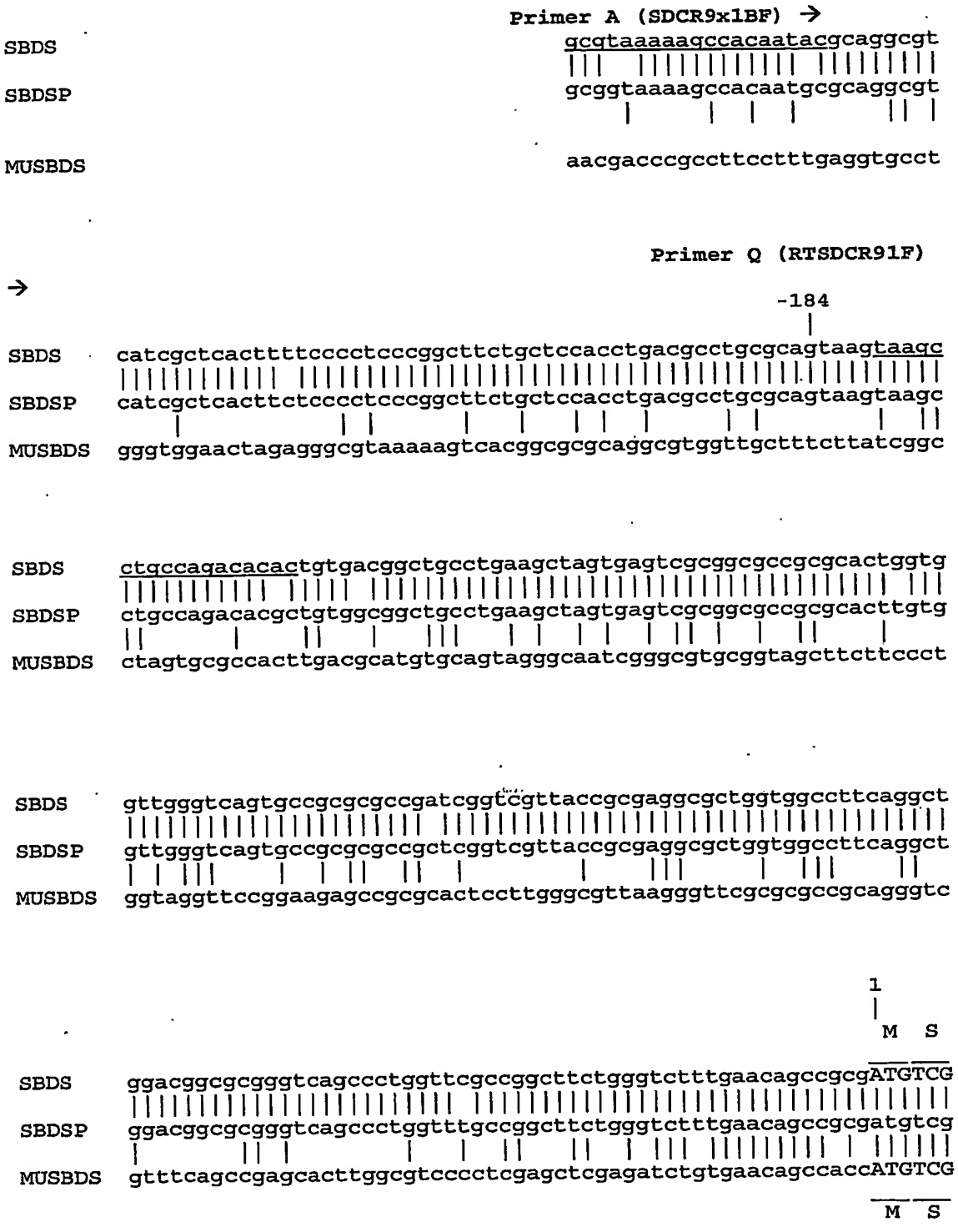
```
 -184 gtaagtaagc ctgccagaca cactgtgacg gctgcctgaa gctagtgagt cgcggcgccg
 -124 cgcactggtg gttgggtcag tgccgcgcgc cgatcggtcg ttaccgcgag gcgctggtgg
  -64 ccttcaggct ggacggcgcg ggtcagccct ggttcgccgg cttctgggtc tttgaacagc
   -4 cgcgATGTCG ATCTTCACCC CCACCAACCA GATCCGCCTA ACCAATGTGG CCGTGGTACG
  +57 GATGAAGCGT GCCGGGAAGC GCTTCGAAAT CGCCTGCTAC AAAAACAAGG TCGTCGGCTG
 +117 GCGGAGCGGC GTGGAAAAAG ACCTCGATGA AGTTCTGCAG ACCCACTCAG TGTTTGTAAA
 +177 TGTTTCTAAA GGTCAGGTTG CCAAAAAGGA AGATCTCATC AGTGCGTTTG GAACAGATGA
 +237 CCAAACTGAA ATCTGTAAGC AGATTTTGAC TAAAGGAGAA GTTCAAGTAT CAGATAAAGA
 +297 AAGACACACA CAACTGGAGC AGATGTTTAG GGACATTGCA ACTATTGTGG CAGACAAATG
 +357 TGTGAATCCT GAAACAAAGA GACCATACAC CGTGATCCTT ATTGAGAGAG CCATGAAGGA
 +417 CATCCACTAT TCGGTGAAAA CCAACAAGAG TACAAAACAG CAGGCTTTGG AAGTGATAAA
 +477 GCAGTTAAAA GAGAAAATGA AGATAGAACG TGCTCACATG AGGCTTCGGT TCATCCTTCC
 +537 AGTCAATGAA GGCAAGAAGC TGAAAGAAAA GCTCAAGCCA CTGATCAAGG TCATAGAAAG
 +597 TGAAGATTAT GGCCAACAGT TAGAAATCGT ATGTCTGATT GACCCGGGCT GCTTCCGAGA
 +657 AATTGATGAG CTAATAAAAA AGGAAACTAA AGGCAAAGGT TCTTTGGAAG TACTCAATCT
 +717 GAAAGATGTA GAAGAAGGAG ATGAGAAATT TGAAtgacac ccatcaatct cttcacctct
 +777 aaaacactaa agtgtttccg tttccgacgg cactgtttca tgtctgtggt ctgccaaata
 +837 cttgcttaaa ctatttgaca ttttctactt tgtgttaaca gtggacacag caaggctttc
 +897 ctacataagt ataataatgt gggaatgatt tggttttaat tataaactgg ggtctaaatc
 +957 ctaaagcaaa attgaaactc caagatgcaa agtccagagt ggcattttgc tactctgtct
+1017 catgccttga tagctttcca aaatgaaagt tacttgaggc agctcttgtg ggtgaaaagt
+1077 tatttgtaca gtagagtaag attattaggg gtatgtctat acaacaaaag ggggggtctt
+1137 tcctaaaaaa gaaaacatat gatgcttcat ttctacttaa tggaacttgt gttctgaggg
+1197 tcattatggt atcgtaatgt aaagcttgga tgatgttcct gattatctga gaaacagata
+1257 tagaaaaatt gtgccggact tacctttcat tgaacatgct gccataactt agattattct
+1317 tggttaaaaa ataaaagtca cttatttcta attcttaaag tttataatat atattaatat
+1397 agctaaaatt gtatgtaatc aataaaacca ctcttatgtt tatt
```

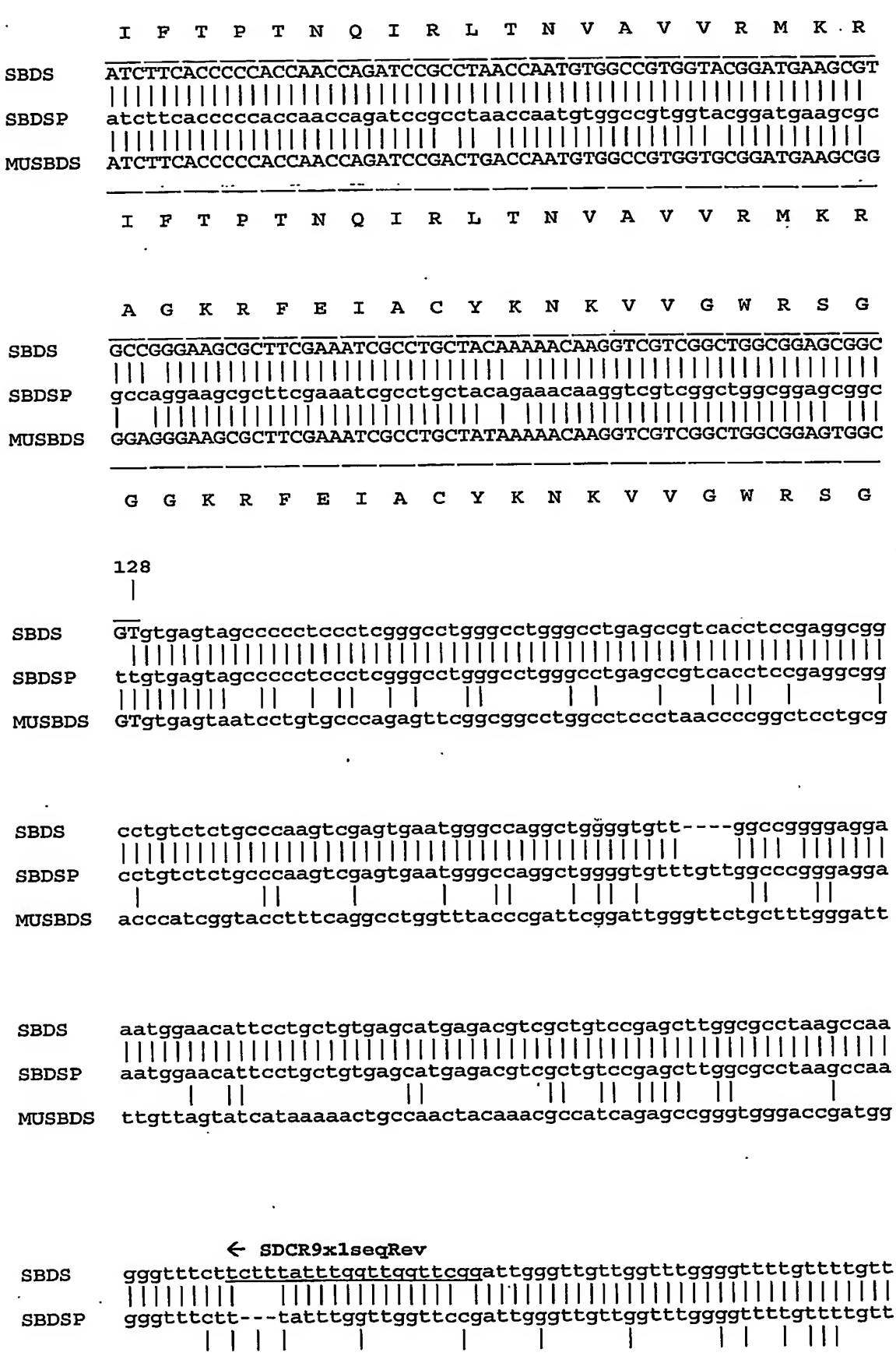
SBDS Amino Acid Sequence ID NO:2

```
  1 MSIFTPTNQI RLTNVAVVRM KRAGKRFEIA CYKNKVVGWR SGVEKDLDEV LQTHSVFVNV
 61 SKGQVAKKED LISAFGTDDQ TEICKQILTK GEVQVSDKER HTQLEQMFRD IATIVADKCV
121 NPETKRPYTV ILIERAMKDI HYSVKTNKST KQQALEVIKQ LKEKMKIERA HMRLRFILPV
181 NEGKKLKEKL KPLIKVIESE DYGQQLEIVC LIDPGCFREI DELIKKETKG KGSLEVLNLK
241 DVEEGDEKFE
```

FIGURE 5

SBDS Exon 1:

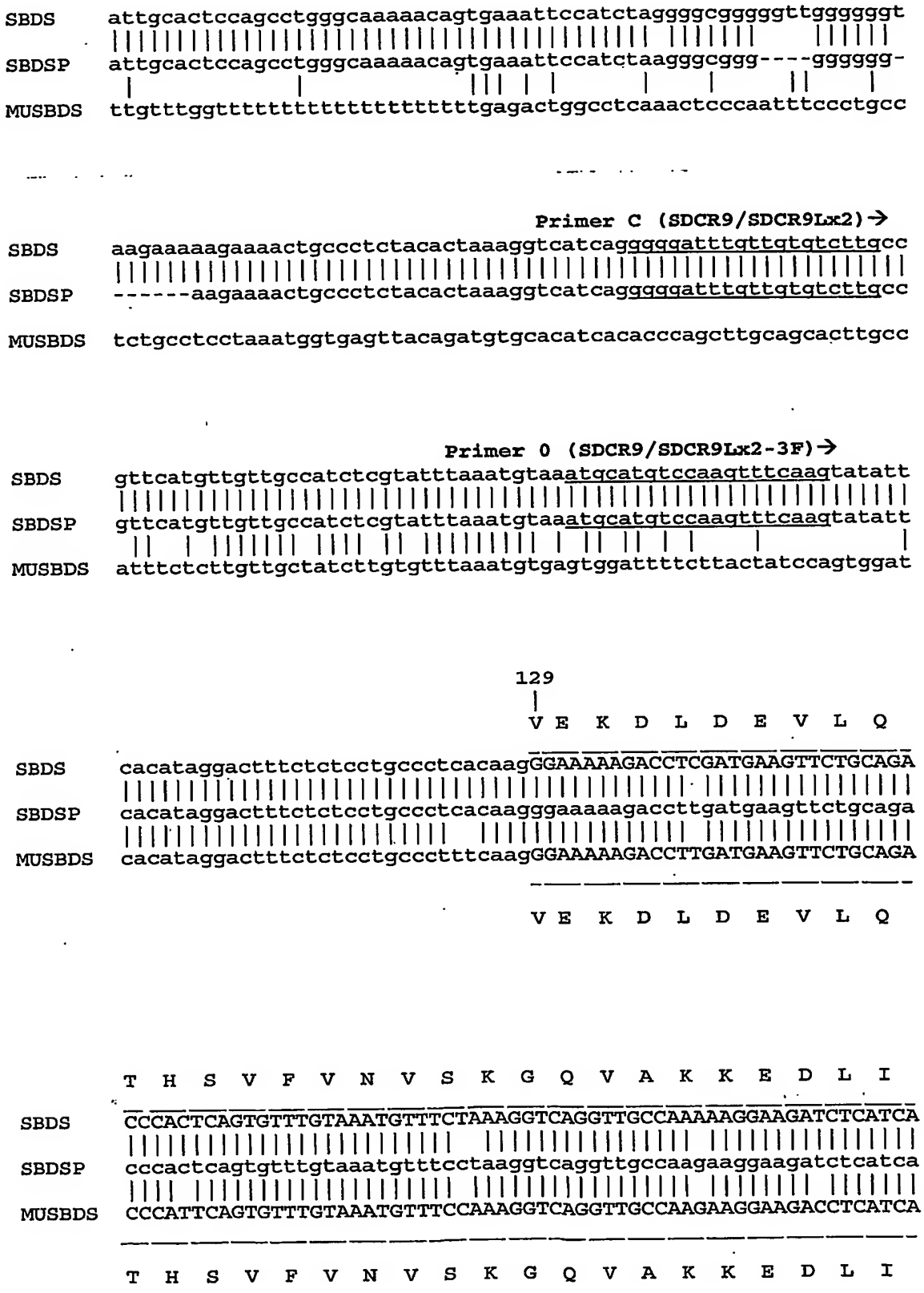

Primer A (SDCR9x1BF) →
SBDS
gcgtaaaaagccacaatacgcaggcgt
SBDSP
gcggtaaaagccacaatgcgcaggcgt
MUSBDS
aacgacccgccttcctttgaggtgcct
Primer Q (RTSDCR91F)
→
-184
SBDS
catcgctcacttttcccctcccggcttctgctccacctgacgcctgcgcagtaagtaagc
SBDSP
catcgctcacttctcccctcccggcttctgctccacctgacgcctgcgcagtaagtaagc
MUSBDS
gggtggaactagagggcgtaaaaagtcacggcgcgcaggcgtggttgctttcttatcggc
SBDS
ctgccagacacactgtgacggctgcctgaagctagtgagtcgcggcgccgcgcactggtg
SBDSP
ctgccagacacgctgtggcggctgcctgaagctagtgagtcgcggcgccgcgcacttgtg
MUSBDS
ctagtgcgccacttgacgcatgtgcagtagggcaatcgggcgtgcggtagcttcttccct
SBDS
gttgggtcagtgccgcgcgccgatcggtcgttaccgcgaggcgctggtggccttcaggct
SBDSP
gttgggtcagtgccgcgcgccgctcggtcgttaccgcgaggcgctggtggccttcaggct
MUSBDS
ggtaggttccggaagagccgcgcactccttgggcgttaagggttcgcgcgccgcagggtc
1
M S
SBDS
ggacggcgcgggtcagccctggttcgccggcttctgggtctttgaacagccgcgATGTCG
SBDSP
ggacggcgcgggtcagccctggtttgccggcttctgggtctttgaacagccgcgatgtcg
MUSBDS
gtttcagccgagcacttggcgtcccctcgagctcgagatctgtgaacagccaccATGTCG
M S

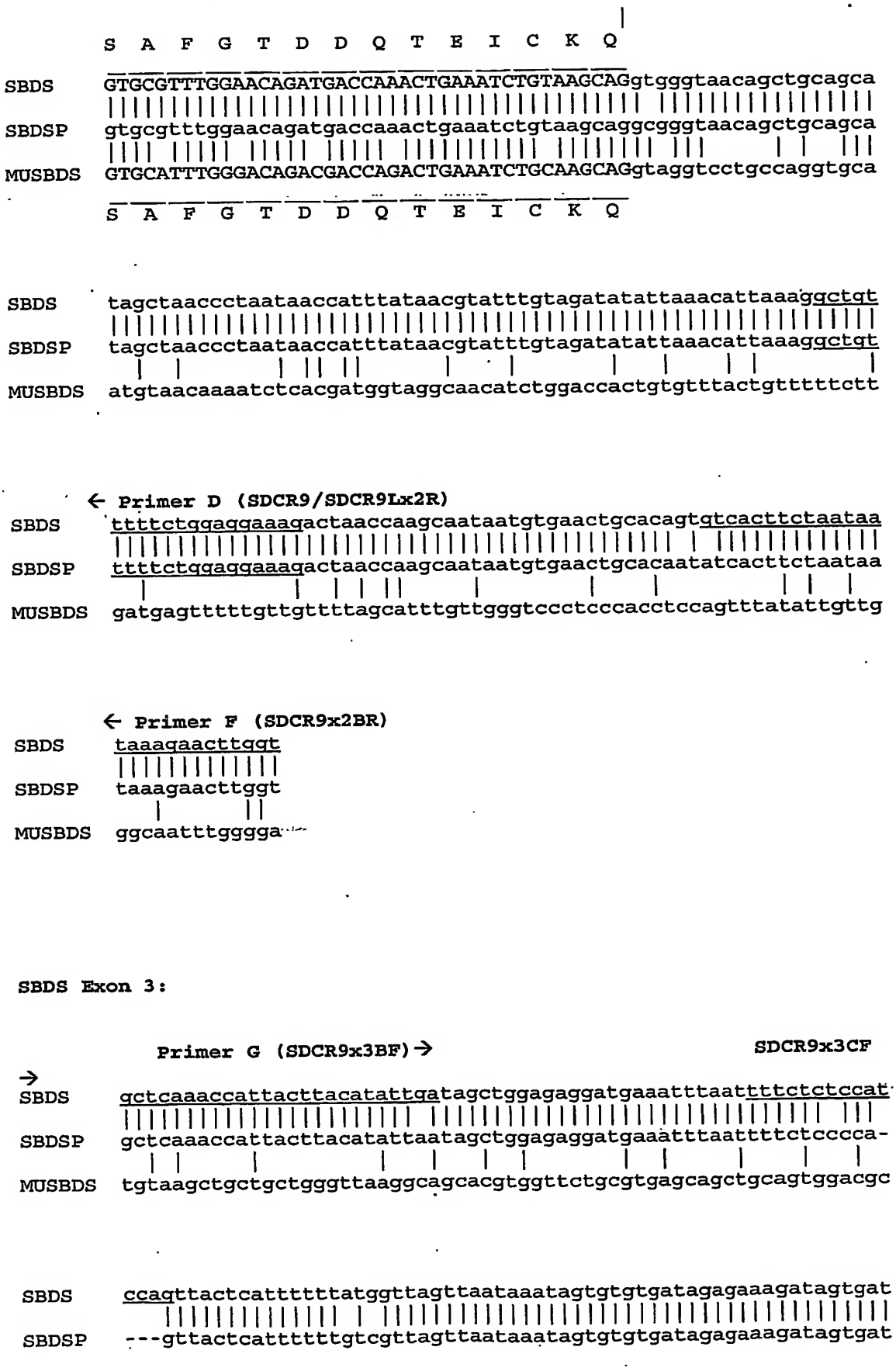

I F T P T N Q I R L T N V A V V R M K R
SBDS ATCTTCACCCCCACCAACCAGATCCGCCTAACCAATGTGGCCGTGGTACGGATGAAGCGT
SBDSP atcttcacccccaccaaccagatccgcctaaccaatgtggccgtggtacggatgaagcgc
MUSBDS ATCTTCACCCCCACCAACCAGATCCGACTGACCAATGTGGCCGTGGTGCGGATGAAGCGG
I F T P T N Q I R L T N V A V V R M K R
A G K R F E I A C Y K N K V V G W R S G
SBDS GCCGGGAAGCGCTTCGAAATCGCCTGCTACAAAAACAAGGTCGTCGGCTGGCGGAGCGGC
SBDSP gccaggaagcgcttcgaaatcgcctgctacagaaacaaggtcgtcggctggcggagcggc
MUSBDS GGAGGGAAGCGCTTCGAAATCGCCTGCTATAAAAACAAGGTCGTCGGCTGGCGGAGTGGC
G G K R F E I A C Y K N K V V G W R S G
128
SBDS GTgtgagtagccccctccctcgggcctgggcctgggcctgagccgtcacctccgaggcgg
SBDSP ttgtgagtagccccctccctcgggcctgggcctgggcctgagccgtcacctccgaggcgg
MUSBDS GTgtgagtaatcctgtgcccagagttcggcggcctggcctccctaaccccggctcctgcg
SBDS cctgtctctgcccaagtcgagtgaatgggccaggctggggtgtt----ggccggggagga
SBDSP cctgtctctgcccaagtcgagtgaatgggccaggctggggtgtttgttggcccgggagga
MUSBDS acccatcggtacctttcaggcctggtttacccgattcggattgggttctgctttgggatt
SBDS aatggaacattcctgctgtgagcatgagacgtcgctgtccgagcttggcgcctaagccaa
SBDSP aatggaacattcctgctgtgagcatgagacgtcgctgtccgagcttggcgcctaagccaa
MUSBDS ttgttagtatcataaaaactgccaactacaaacgccatcagagccgggtgggaccgatgg
← SDCR9x1seqRev
SBDS gggtttcttctttatttggttggttcggattgggttgttggtttggggttttgttttgtt
SBDSP gggtttctt---tatttggttggttccgattgggttgttggtttggggttttgttttgtt

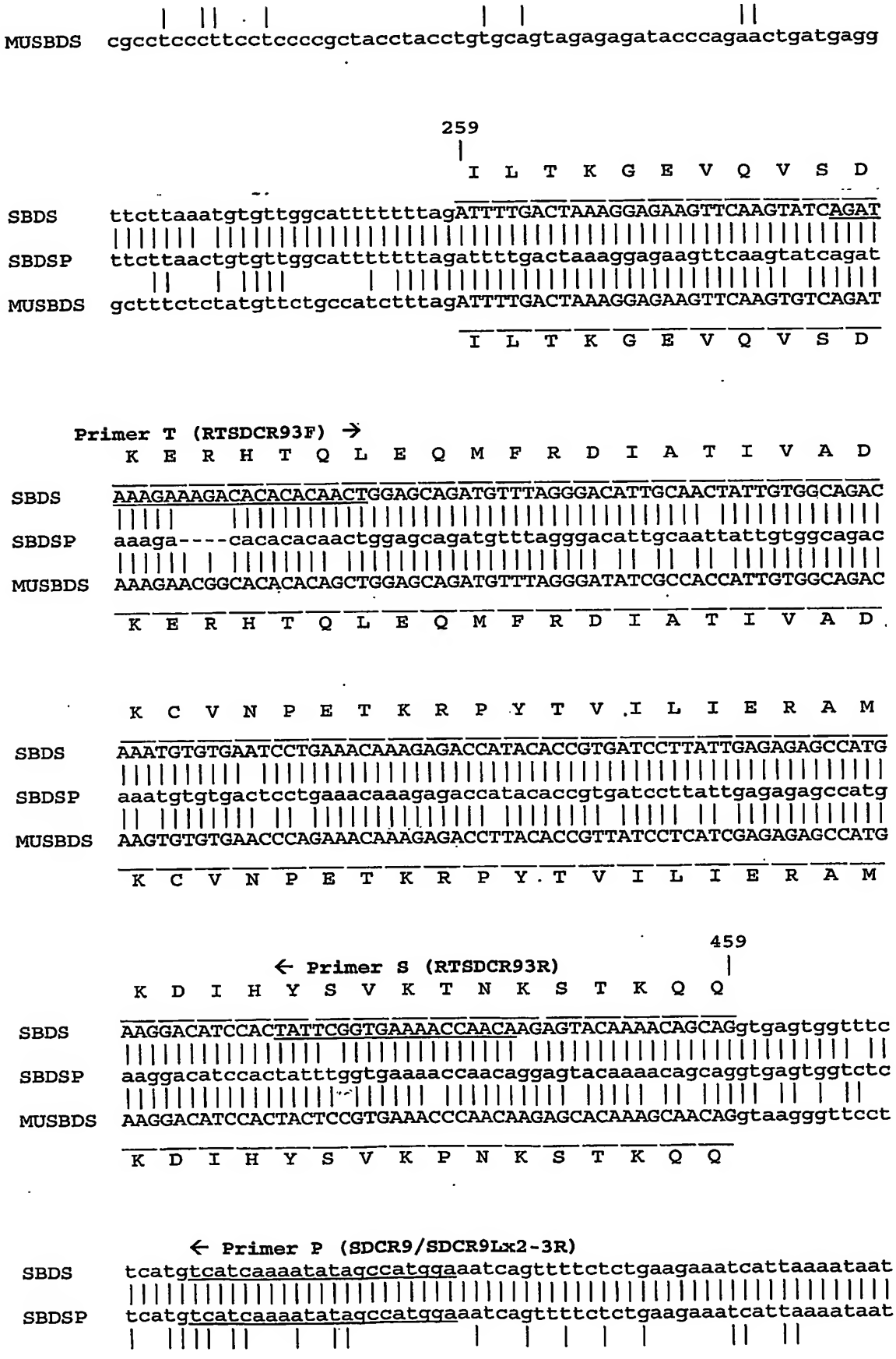
```
MUSBDS  tttaggcctgtaatcccagcgcccaggaaactgaggcaggaggattgctgcgatttccag

SBDS    ggtgtcataaaagctgcagccaagaaatctcgtaattgtggtccttttcctagaataatg
        ||||||||||||||||||||||||||||||| ||||||||||||||||||||||||||||
SBDSP   ggtgtcataaaagctgcagccaagaaatctcataattgtggtccttttcctagaataatg
        |    | |  ||  ||       |      | ||  ||||     |        ||
MUSBDS  gccagcctggaacgtgtgtgtgtgtgtatgtgtatgtgtgtgttgtgtgtgtgtatgtgt

                    ← Primer B (SDCR9x1BR)
SBDS    atggctgagaacctagtcttacgaatactgtcatag
        ||||||||||||||||  ||  ||||||||||||||
SBDSP   atggctgagaacctagtgttccgaatactgtcatag
        |||  ||  |             ||         |
MUSBDS  atgtgtgtgtgagagagaccgtgaccgaccctgtac
```

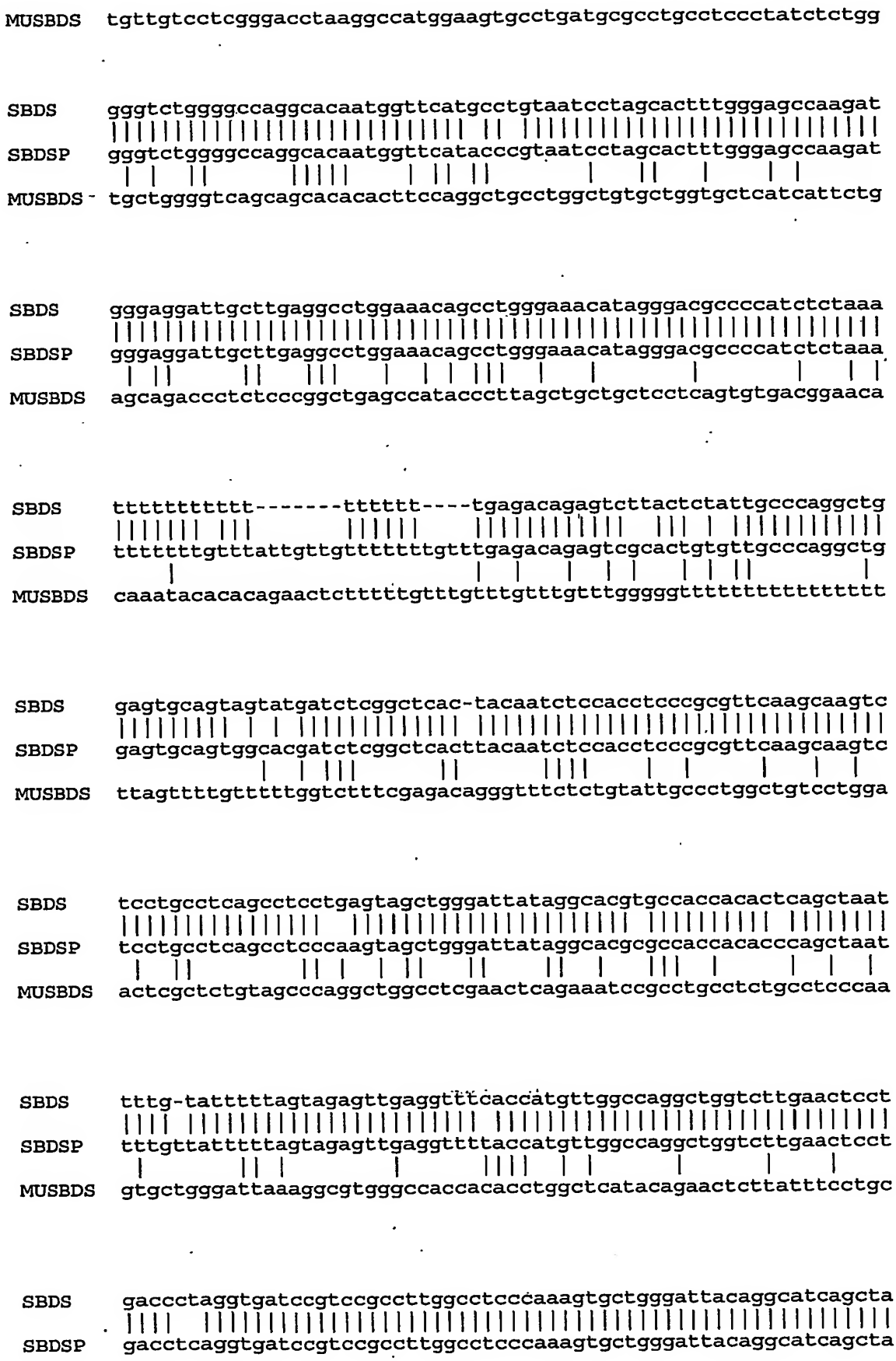
SBDS Exon 2:

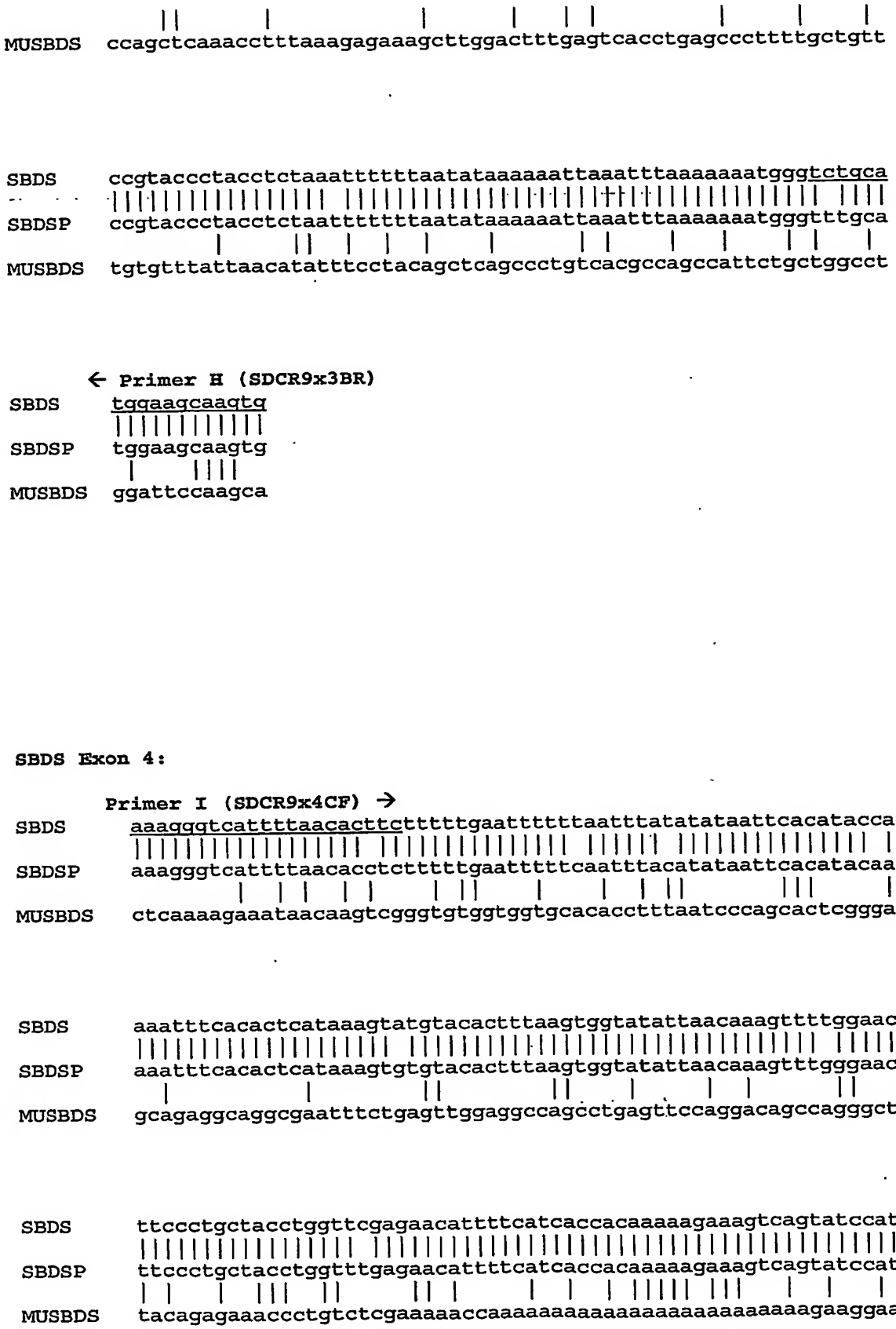
```
      Primer E (SDCR9x2BF)→
SBDS    aaatggtaaggcaaatacggttctgagttttgaaaatgttccctcaggccgatgcgggca
        |||||||| ||||||||| ||||||||||||||||||||||||||||||||||||||||
SBDSP   aaatggtagggcaaatacagttctgagttttgaaaatgttccctcaggccgatgcgggca
          |  ||       |      |||  |    ||  |  |              |   |
MUSBDS  gtagtgtcttcgctactgccatctagggacagatattccaggacagaagaaacaccactc

SBDS    gttcacttgaggccaggagttcgaggccagcctggccaacatgaaaccccatctctacta
        | ||||||||||||||||||||||||||||||||||||||||||||| ||||||||||||
SBDSP   gatcacttgaggccaggagttcgaggccagcctggccaacatgaaacaccatctctacta
           |        ||       |    |  |        |      |||         |
MUSBDS  cccaccacaccctgagtttccttacataaaacaatgatgtagtttttccctctgtggtga

SBDS    aaaatacaaagttagccgggtgtggtggcgcatgcctgtaatcccagttactcaggaggc
        |||||||||| ||||||||||||||||||||||||||||||||||||| ||||||||||||
SBDSP   aaaatacaaaattagccgggtgtggtggcgcatgcctgtaatcccagctactcaggaggc
        |       ||   ||         | ||   |  |     |          |   |
MUSBDS  agtgggagaatccagatactgtccttcgcaggtagccaccagagagagagtgtggtgtgt

SBDS    tgaggcgggagaatcacttgaacccgggaggctgaggttacagtgacccgagatcgcgcc
        ||||||| ||||||||||||||||||||||| || ||| |||||| ||||||||||||||
SBDSP   tgaggcaggagaatcacttgaacccgggaggcggacgttgcagtgagccgagatcgcgcc
           | | ||||| |    |                |  ||||   ||        |
MUSBDS  gtgtgtgtgagatttctctttttttttttctttagggtttttgttttgttttttttgtt
```

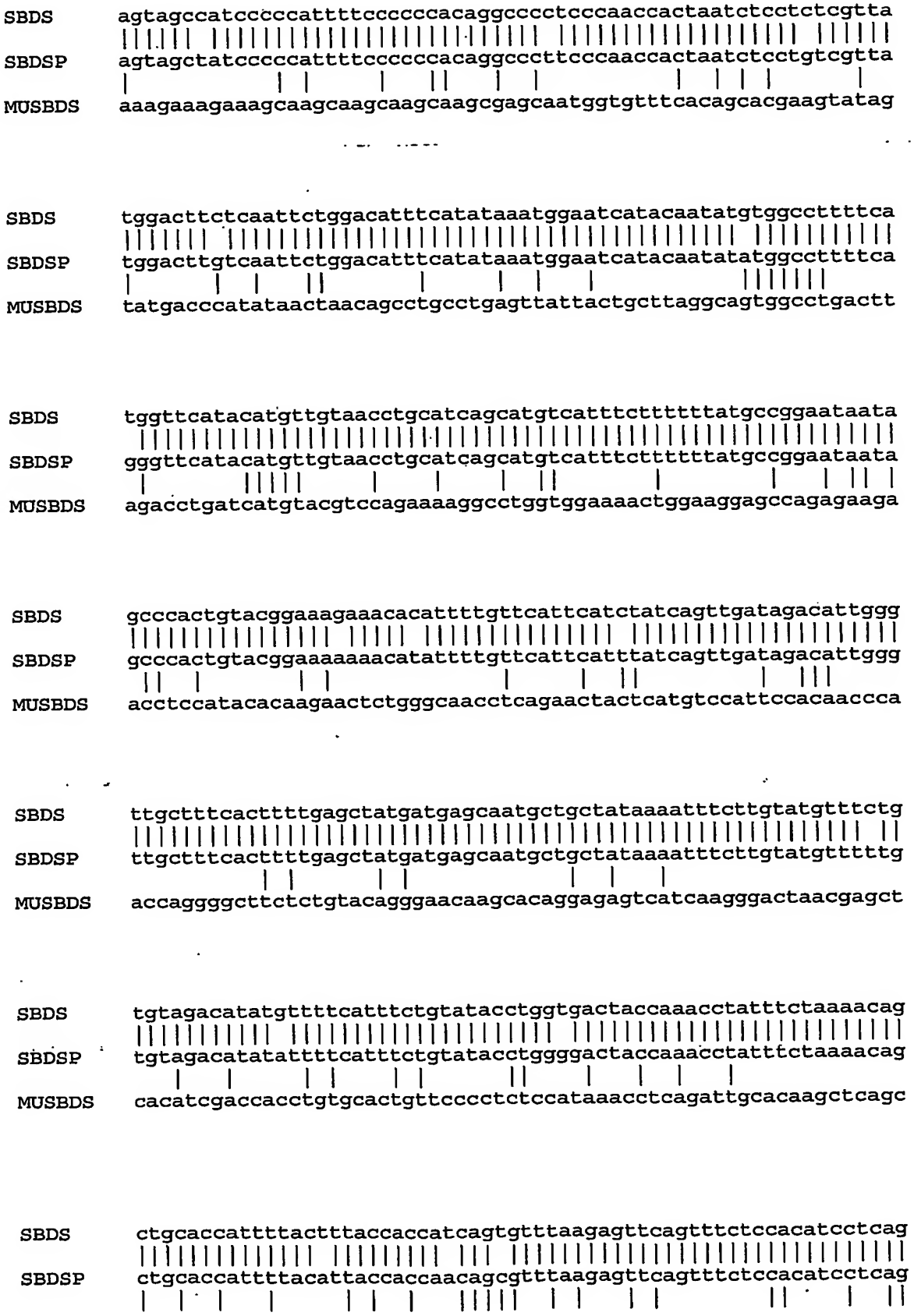

SBDS attgcactccagcctgggcaaaaacagtgaaattccatctaggggcgggggttgggggt
SBDSP attgcactccagcctgggcaaaaacagtgaaattccatctaagggcggg----gggggg-
MUSBDS ttgtttggtttttttttttttttttttgagactggcctcaaactcccaatttccctgcc
Primer C (SDCR9/SDCR9Lx2)→
SBDS aagaaaaagaaaactgccctctacactaaaggtcatcaggggggatttgttgtgtcttgcc
SBDSP ------aagaaaactgccctctacactaaaggtcatcaggggggatttgttgtgtcttgcc
MUSBDS tctgcctcctaaatggtgagttacagatgtgcacatcacacccagcttgcagcacttgcc
Primer 0 (SDCR9/SDCR9Lx2-3F)→
SBDS gttcatgttgttgccatctcgtatttaaatgtaaatgcatgtccaagtttcaagtatatt
SBDSP gttcatgttgttgccatctcgtatttaaatgtaaatgcatgtccaagtttcaagtatatt
MUSBDS atttctcttgttgctatcttgtgtttaaatgtgagtggattttcttactatccagtggat
129
V E K D L D E V L Q
SBDS cacataggactttctctcctgccctcacaagGGAAAAAGACCTCGATGAAGTTCTGCAGA
SBDSP cacataggactttctctcctgccctcacaagggaaaaagaccttgatgaagttctgcaga
MUSBDS cacataggactttctctcctgcccttcaagGGAAAAAGACCTTGATGAAGTTCTGCAGA
V E K D L D E V L Q
T H S V F V N V S K G Q V A K K E D L I
SBDS CCCACTCAGTGTTTGTAAATGTTTCTAAAGGTCAGGTTGCCAAAAAGGAAGATCTCATCA
SBDSP cccactcagtgtttgtaaatgtttcctaaggtcaggttgccaagaaggaagatctcatca
MUSBDS CCCATTCAGTGTTTGTAAATGTTTCCAAAGGTCAGGTTGCCAAGAAGGAAGACCTCATCA
T H S V F V N V S K G Q V A K K E D L I

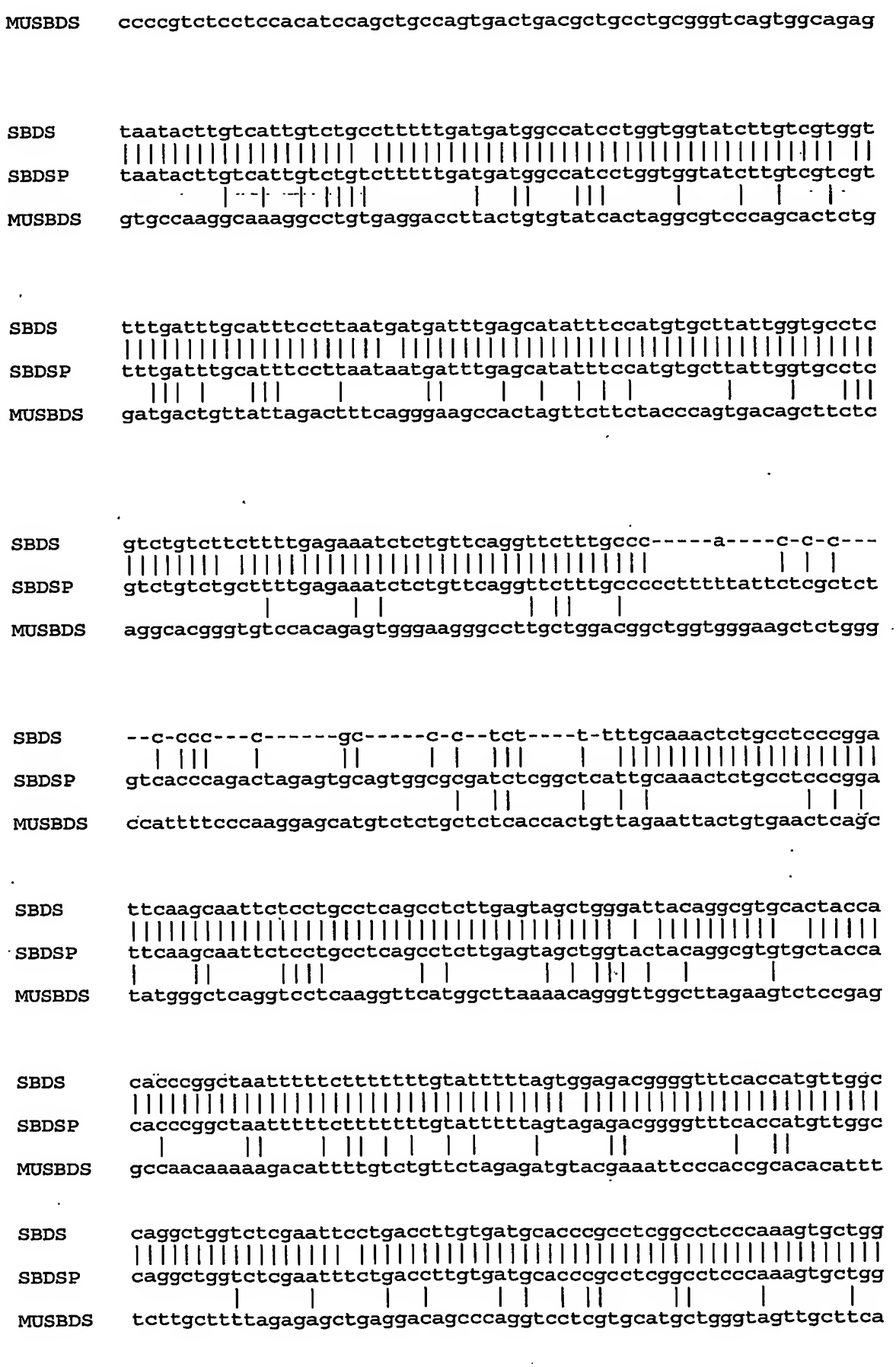

S A F G T D D Q T E I C K Q
SBDS GTGCGTTTGGAACAGATGACCAAACTGAAATCTGTAAGCAGgtgggtaacagctgcagca
SBDSP gtgcgtttggaacagatgaccaaactgaaatctgtaagcaggcgggtaacagctgcagca
MUSBDS GTGCATTTGGGACAGACGACCAGACTGAAATCTGCAAGCAGgtaggtcctgccaggtgca
S A F G T D D Q T E I C K Q
SBDS tagctaaccctaataaccatttataacgtatttgtagatatattaaacattaaaggctgt
SBDSP tagctaaccctaataaccatttataacgtatttgtagatatattaaacattaaaggctgt
MUSBDS atgtaacaaaatctcacgatggtaggcaacatctggaccactgtgtttactgtttttctt
← Primer D (SDCR9/SDCR9Lx2R)
SBDS ttttctggaggaaagactaaccaagcaataatgtgaactgcacagtgtcacttctaataa
SBDSP ttttctggaggaaagactaaccaagcaataatgtgaactgcacaatatcacttctaataa
MUSBDS gatgagttttgttgttttagcatttgttgggtccctcccacctccagtttatattgttg
← Primer F (SDCR9x2BR)
SBDS taaagaacttggt
SBDSP taaagaacttggt
MUSBDS ggcaatttgggga
SBDS Exon 3:
Primer G (SDCR9x3BF)→
SDCR9x3CF
→
SBDS gctcaaaccattacttacatattgatagctggagaggatgaaatttaattttctctccat
SBDSP gctcaaaccattacttacatattaatagctggagaggatgaaatttaattttctcccca-
MUSBDS tgtaagctgctgctgggttaaggcagcacgtggttctgcgtgagcagctgcagtggacgc
SBDS ccagttactcatttttatggttagttaataaatagtgtgtgatagagaaagatagtgat
SBDSP ---gttactcatttttgtcgttagttaataaatagtgtgtgatagagaaagatagtgat

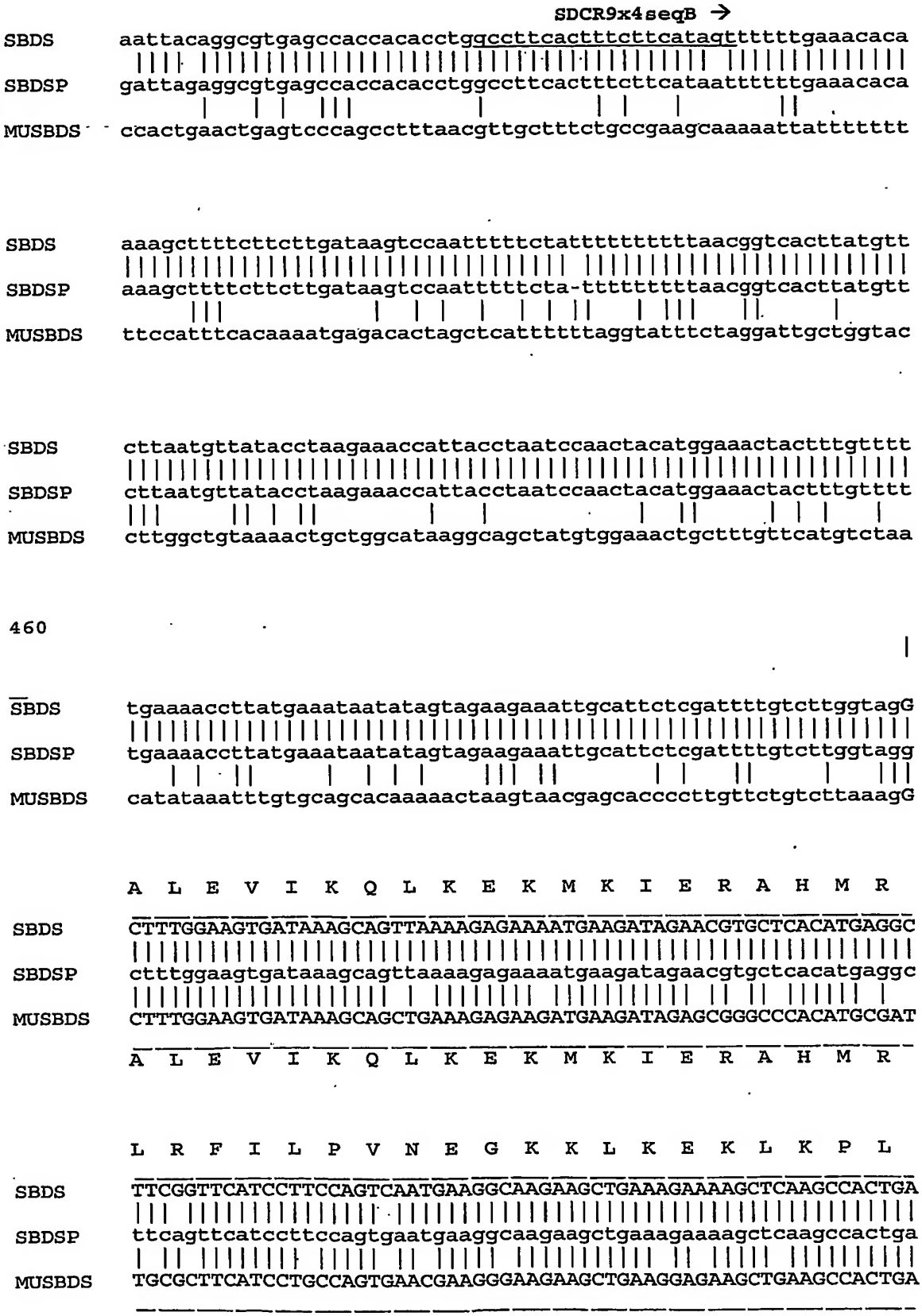

MUSBDS cgcctcccttcctccccgctacctacctgtgcagtagagagatacccagaactgatgagg
259
I L T K G E V Q V S D
SBDS ttcttaaatgtgttggcatttttttagATTTTGACTAAAGGAGAAGTTCAAGTATCAGAT
SBDSP ttcttaactgtgttggcatttttttagattttgactaaaggagaagttcaagtatcagat
MUSBDS gctttctctatgttctgccatctttagATTTTGACTAAAGGAGAAGTTCAAGTGTCAGAT
I L T K G E V Q V S D
Primer T (RTSDCR93F) →
K E R H T Q L E Q M F R D I A T I V A D
SBDS AAAGAAAGACACACACAACTGGAGCAGATGTTTAGGGACATTGCAACTATTGTGGCAGAC
SBDSP aaaga----cacacacaactggagcagatgtttagggacattgcaattattgtggcagac
MUSBDS AAAGAACGGCACACACAGCTGGAGCAGATGTTTAGGGATATCGCCACCATTGTGGCAGAC
K E R H T Q L E Q M F R D I A T I V A D
K C V N P E T K R P Y T V I L I E R A M
SBDS AAATGTGTGAATCCTGAAACAAAGAGACCATACACCGTGATCCTTATTGAGAGAGCCATG
SBDSP aaatgtgtgactcctgaaacaaagagaccatacaccgtgatccttattgagagagccatg
MUSBDS AAGTGTGTGAACCCAGAAACAAAGAGACCTTACACCGTTATCCTCATCGAGAGAGCCATG
K C V N P E T K R P Y T V I L I E R A M
459
← Primer S (RTSDCR93R)
K D I H Y S V K T N K S T K Q Q
SBDS AAGGACATCCACTATTCGGTGAAAACCAACAAGAGTACAAAACAGCAGgtgagtggtttc
SBDSP aaggacatccactatttggtgaaaaccaacaggagtacaaaacagcaggtgagtggtctc
MUSBDS AAGGACATCCACTACTCCGTGAAACCCAACAAGAGCACAAAGCAACAGgtaagggttcct
K D I H Y S V K P N K S T K Q Q
← Primer P (SDCR9/SDCR9Lx2-3R)
SBDS tcatgtcatcaaaatatagccatggaaatcagttttctctgaagaaatcattaaaataat
SBDSP tcatgtcatcaaaatatagccatggaaatcagttttctctgaagaaatcattaaaataat


MUSBDS tgttgtcctcgggacctaaggccatggaagtgcctgatgcgcctgcctccctatctctgg
SBDS gggtctggggccaggcacaatggttcatgcctgtaatcctagcactttgggagccaagat
SBDSP gggtctggggccaggcacaatggttcatacccgtaatcctagcactttgggagccaagat
MUSBDS tgctggggtcagcagcacacacttccaggctgcctggctgtgctggtgctcatcattctg
SBDS gggaggattgcttgaggcctggaaacagcctgggaaacatagggacgccccatctctaaa
SBDSP gggaggattgcttgaggcctggaaacagcctgggaaacatagggacgccccatctctaaa
MUSBDS agcagaccctctcccggctgagccataccttagctgctgctcctcagtgtgacggaaca
SBDS ttttttttttt-------tttttt----tgagacagagtcttactctattgcccaggctg
SBDSP ttttttttgtttattgttgttttttttgtttgagacagagtcgcactgtgttgcccaggctg
MUSBDS caaatacacacagaactctttttgtttgtttgtttgtttgggggttttttttttttttttt
SBDS gagtgcagtagtatgatctcggctcac-tacaatctccacctcccgcgttcaagcaagtc
SBDSP gagtgcagtggcacgatctcggctcacttacaatctccacctcccgcgttcaagcaagtc
MUSBDS ttagttttgtttttggtctttcgagacagggtttctctgtattgccctggctgtcctgga
SBDS tcctgcctcagcctcctgagtagctgggattataggcacgtgccaccacactcagctaat
SBDSP tcctgcctcagcctcccaagtagctgggattataggcacgcgccaccacacccagctaat
MUSBDS actcgctctgtagcccaggctggcctcgaactcagaaatccgcctgcctctgcctcccaa
SBDS tttg-tatttttagtagagttgaggtttcaccatgttggccaggctggtcttgaactcct
SBDSP tttgttatttttagtagagttgaggttttaccatgttggccaggctggtcttgaactcct
MUSBDS gtgctgggattaaaggcgtgggccaccacacctggctcatacagaactcttatttcctgc
SBDS gaccctaggtgatccgtccgccttggcctcccaaagtgctgggattacaggcatcagcta
SBDSP gacctcaggtgatccgtccgccttggcctcccaaagtgctgggattacaggcatcagcta


MUSBDS ccagctcaaacctttaaagagaaagcttggactttgagtcacctgagccctttttgctgtt
SBDS ccgtaccctacctctaaatttttttaatataaaaaattaaatttaaaaaaatgggtctgca
SBDSP ccgtaccctacctctaatttttttaatataaaaaattaaatttaaaaaaatgggtttgca
MUSBDS tgtgtttattaacatatttcctacagctcagccctgtcacgccagccattctgctggcct
← Primer H (SDCR9x3BR)
SBDS tggaagcaagtg
SBDSP tggaagcaagtg
MUSBDS ggattccaagca
SBDS Exon 4:
Primer I (SDCR9x4CF) →
SBDS aaagggtcattttaacacttcttttttgaatttttttaatttatatataattcacataccat
SBDSP aaagggtcattttaacacctctttttgaatttttcaatttacatataattcacatacaat
MUSBDS ctcaaaagaaataacaagtcgggtgtggtggtgcacacctttaatcccagcactcgggag
SBDS aaatttcacactcataaagtatgtacactttaagtggtatattaacaaagttttggaacc
SBDSP aaatttcacactcataaagtgtgtacactttaagtggtatattaacaaagtttgggaacc
MUSBDS gcagaggcaggcgaatttctgagttggaggccagcctgagttccaggacagccagggcta
SBDS ttccctgctacctggttcgagaacattttcatcaccacaaaaagaaagtcagtatccatt
SBDSP ttccctgctacctggtttgagaacattttcatcaccacaaaaagaaagtcagtatccatt
MUSBDS tacagagaaaccctgtctcgaaaaaccaaaaaaaaaaaaaaaaaaaaaaaaagaaggaag


SBDS agtagccatcccccatttcccccccacaggcccctcccaaccactaatctcctctcgtta
SBDSP agtagctatcccccatttcccccccacaggcccttcccaaccactaatctcctgtcgtta
MUSBDS aaagaaagaaagcaagcaagcaagcaagcgagcaatggtgtttcacagcacgaagtatag
SBDS tggacttctcaattctggacatttcatataaatggaatcatacaatatgtggccttttca
SBDSP tggacttgtcaattctggacatttcatataaatggaatcatacaatatatggccttttca
MUSBDS tatgacccatataactaacagcctgcctgagttattactgcttaggcagtggcctgactt
SBDS tggttcatacatgttgtaacctgcatcagcatgtcatttctttttttatgccggaataata
SBDSP gggttcatacatgttgtaacctgcatcagcatgtcatttctttttttatgccggaataata
MUSBDS agacctgatcatgtacgtccagaaaaggcctggtggaaaactggaaggagccagagaaga
SBDS gcccactgtacggaaagaaacacattttgttcattcatctatcagttgatagacattggg
SBDSP gcccactgtacggaaaaaaacatattttgttcattcatttatcagttgatagacattggg
MUSBDS acctccatacacaagaactctgggcaacctcagaactactcatgtccattccacaaccca
SBDS ttgctttcacttttgagctatgatgagcaatgctgctataaaatttcttgtatgtttctg
SBDSP ttgctttcacttttgagctatgatgagcaatgctgctataaaatttcttgtatgtttttg
MUSBDS accaggggcttctctgtacagggaacaagcacaggagagtcatcaagggactaacgagct
SBDS tgtagacatatgttttcatttctgtatacctggtgactaccaaacctatttctaaaacag
SBDSP tgtagacatatattttcatttctgtatacctggggactaccaaacctatttctaaaacag
MUSBDS cacatcgaccacctgtgcactgttcccctctccataaacctcagattgcacaagctcagc
SBDS ctgcaccattttactttaccaccatcagtgtttaagagttcagtttctccacatcctcag
SBDSP ctgcaccattttacattaccaccaacagcgtttaagagttcagtttctccacatcctcag

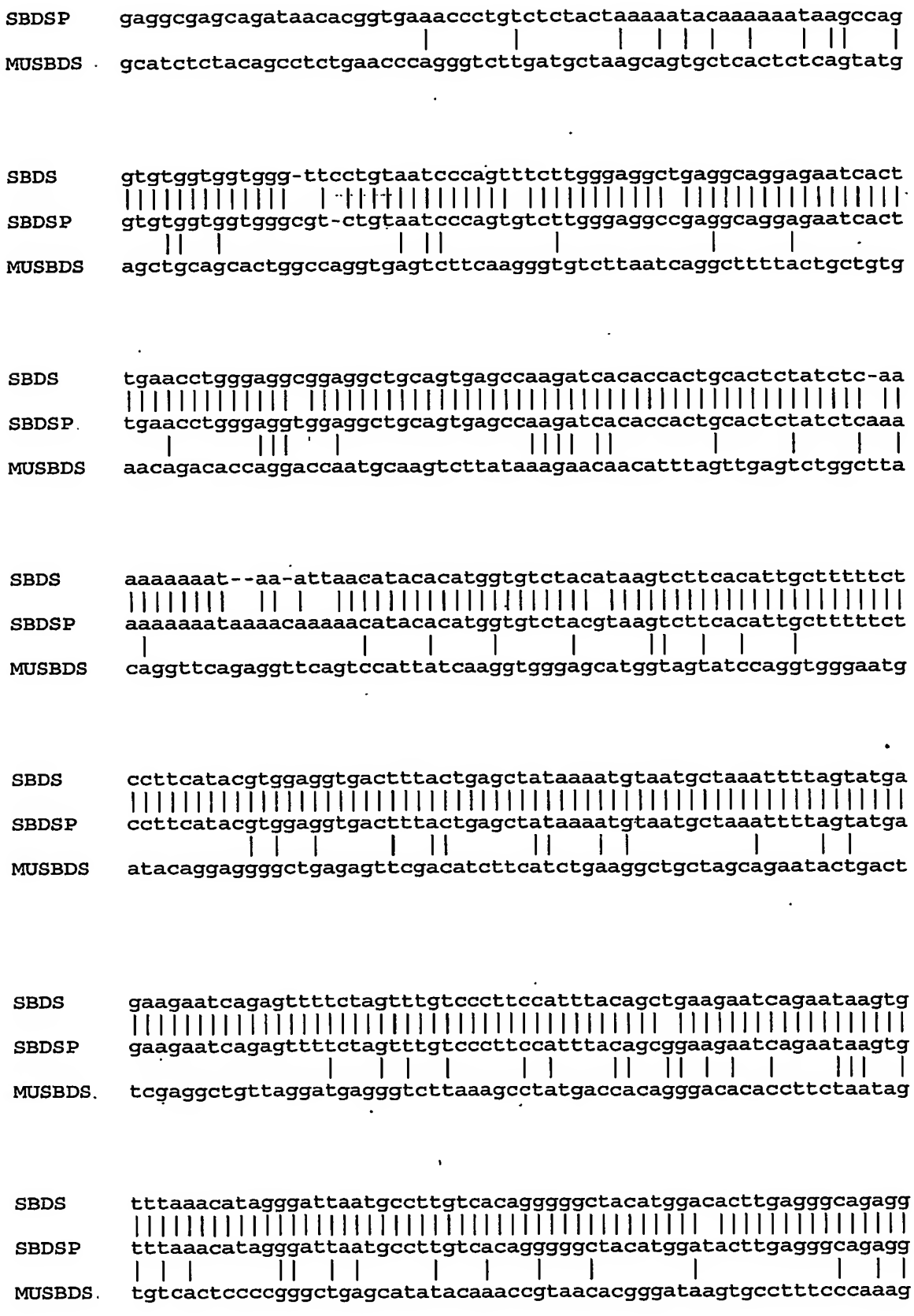

MUSBDS ccccgtctcctccacatccagctgccagtgactgacgctgcctgcgggtcagtggcagag
SBDS taatacttgtcattgtctgccttttgatgatggccatcctggtggtatcttgtcgtggt
SBDSP taatacttgtcattgtctgtcttttgatgatggccatcctggtggtatcttgtcgtcgt
MUSBDS gtgccaaggcaaaggcctgtgaggaccttactgtgtatcactaggcgtcccagcactctg
SBDS tttgatttgcatttccttaatgatgatttgagcatatttccatgtgcttattggtgcctc
SBDSP tttgatttgcatttccttaataatgatttgagcatatttccatgtgcttattggtgcctc
MUSBDS gatgactgttattagactttcagggaagccactagttcttctacccagtgacagcttctc
SBDS gtctgtcttcttttgagaaatctctgttcaggttctttgccc-----a----c-c-c---
SBDSP gtctgtctgcttttgagaaatctctgttcaggttctttgccccctttttattctcgctct
MUSBDS aggcacgggtgtccacagagtgggaagggccttgctggacggctggtgggaagctctggg
SBDS --c-ccc---c------gc-----c-c--tct----t-tttgcaaactctgcctcccgga
SBDSP gtcacccagactagagtgcagtggcgcgatctcggctcattgcaaactctgcctcccgga
MUSBDS ccattttcccaaggagcatgtctctgctctcaccactgttagaattactgtgaactcagc
SBDS ttcaagcaattctcctgcctcagcctcttgagtagctgggattacaggcgtgcactacca
SBDSP ttcaagcaattctcctgcctcagcctcttgagtagctggtactacaggcgtgtgctacca
MUSBDS tatgggctcaggtcctcaaggttcatggcttaaaacagggttggcttagaagtctccgag
SBDS cacccggctaatttttctttttttgtatttttagtggagacggggtttcaccatgttggc
SBDSP cacccggctaatttttctttttttgtatttttagtagagacggggtttcaccatgttggc
MUSBDS gccaacaaaaagacattttgtctgttctagagatgtacgaaattcccaccgcacacattt
SBDS caggctggtctcgaattcctgaccttgtgatgcacccgcctcggcctcccaaagtgctgg
SBDSP caggctggtctcgaatttctgaccttgtgatgcacccgcctcggcctcccaaagtgctgg
MUSBDS tcttgcttttagagagctgaggacagcccaggtcctcgtgcatgctgggtagttgcttca

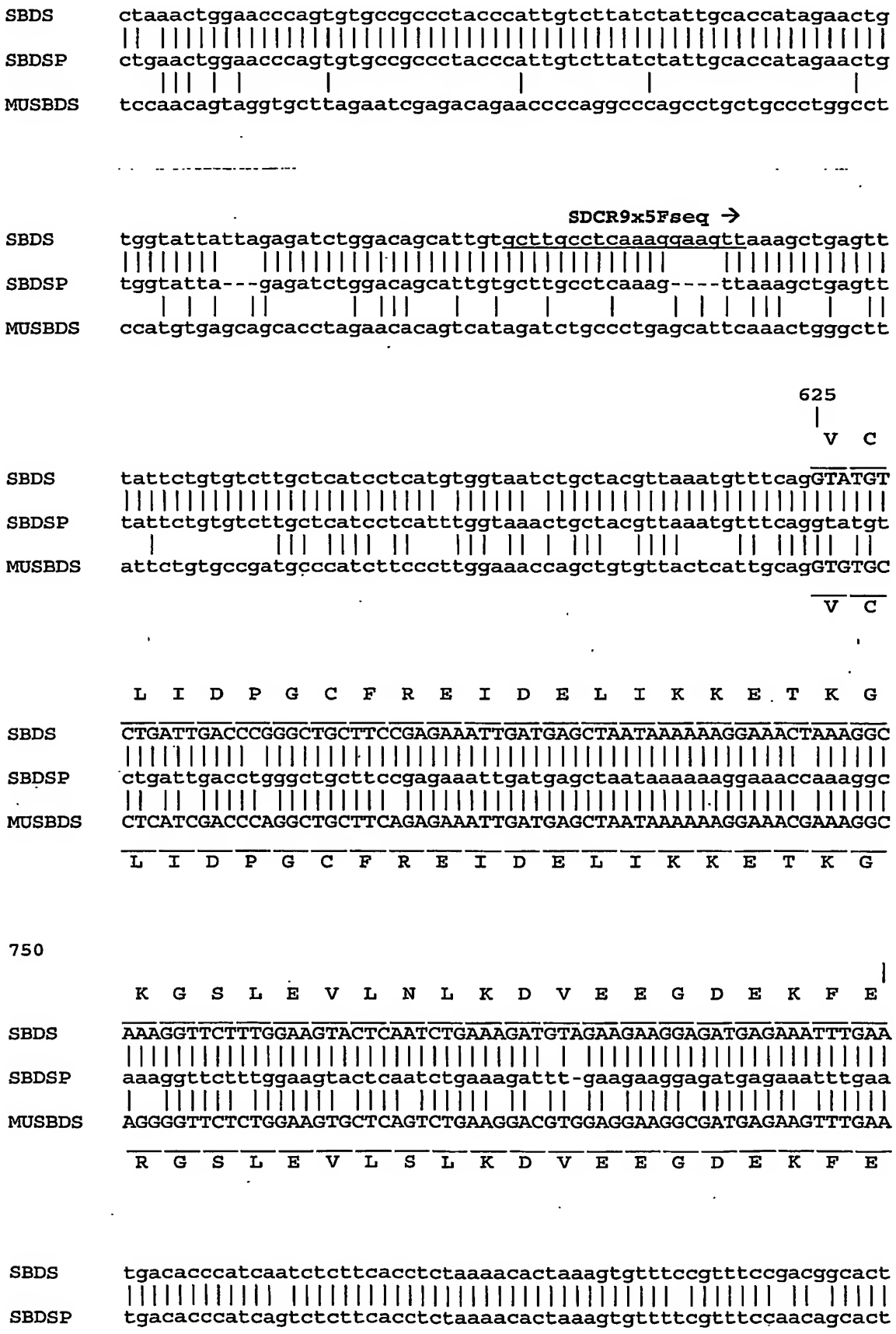

SDCR9x4seqB →
SBDS aattacaggcgtgagccaccacacctggccttcactttcttcatagttttttgaaacaca
SBDSP gattagaggcgtgagccaccacacctggccttcactttcttcataatttttgaaacaca
MUSBDS ccactgaactgagtcccagcctttaacgttgctttctgccgaagcaaaaattatttttt
SBDS aaagcttttcttcttgataagtccaatttttctatttttttttttaacggtcacttatgtt
SBDSP aaagcttttcttcttgataagtccaatttttcta-ttttttttttaacggtcacttatgtt
MUSBDS ttccatttcacaaaatgagacactagctcatttttaggtatttctaggattgctggtac
SBDS cttaatgttatacctaagaaaccattacctaatccaactacatggaaactactttgtttt
SBDSP cttaatgttatacctaagaaaccattacctaatccaactacatggaaactactttgtttt
MUSBDS cttggctgtaaaactgctggcataaggcagctatgtggaaactgctttgttcatgtctaa
460
SBDS tgaaaaccttatgaaataatatagtagaagaaattgcattctcgattttgtcttggtagG
SBDSP tgaaaaccttatgaaataatatagtagaagaaattgcattctcgattttgtcttggtagg
MUSBDS catataaatttgtgcagcacaaaaactaagtaacgagcacccttgttctgtcttaaagG
A L E V I K Q L K E K M K I E R A H M R
SBDS CTTTGGAAGTGATAAAGCAGTTAAAAGAGAAAATGAAGATAGAACGTGCTCACATGAGGC
SBDSP ctttggaagtgataaagcagttaaaagagaaaatgaagatagaacgtgctcacatgaggc
MUSBDS CTTTGGAAGTGATAAAGCAGCTGAAAGAGAAGATGAAGATAGAGCGGGCCCACATGCGAT
A L E V I K Q L K E K M K I E R A H M R
L R F I L P V N E G K K L K E K L K P L
SBDS TTCGGTTCATCCTTCCAGTCAATGAAGGCAAGAAGCTGAAAGAAAAGCTCAAGCCACTGA
SBDSP ttcagttcatccttccagtgaatgaaggcaagaagctgaaagaaaagctcaagccactga
MUSBDS TGCGCTTCATCCTGCCAGTGAACGAAGGGAAGAAGCTGAAGGAGAAGCTGAAGCCACTGA

```
                L  R  F  I  L  P  V  N  E  G  K  K  L  K  E  K  L  K  P  L

                                                                  624
                                                                   |
                I  K  V  I  E  S  E  D  Y  G  Q  Q  L  E  I
                ___________________________________________
SBDS            TCAAGGTCATAGAAAGTGAAGATTATGGCCAACAGTTAGAAATCgtaagagtcaaatatt
                |||||||||||||||||| |||||||||||||||||||||||||||||||||||||||
SBDSP           tcaaggtcatagaaagtaaagattatggccaacagttagaaatcgtaagagtcaaatatt
                | |||||  | ||  ||||| ||  ||  |||| |||  | ||  |||||||||      |
MUSBDS          TGAAGGTGGTGGAGAGTGAGGACTACAGCCAGCAGCTGGAGATCgtaagatgatggtggc
                ___________________________________________
                M  K  V  V  E  S  E  D  Y  S  Q  Q  L  E  I


SBDS            ttctttgcttcatgttacctaaatattgtattctctagtaataaatttgtagcaaacatt
                ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
SBDSP           ttctttgcttcatgttacctaaatattgtattctctagtaataaatttgtagcaaacatt
                                  | | ||           |       |||     ||  |   ||
MUSBDS          ggggagcaggtggcgcagccaaggtcccatgattatgaccttaacacattattattcttg


                    ← Primer J (SDCR9x4CR)
SBDS            tagatgttgtaaac-gtcagatattttc
                 |||  |||||||| ||||||||||||||
SBDSP           cagacattgtaaacagtcagatattttc
                      || ||  |    ||     |||
MUSBDS          gcttccttctacccaaatagcctcgttc
```

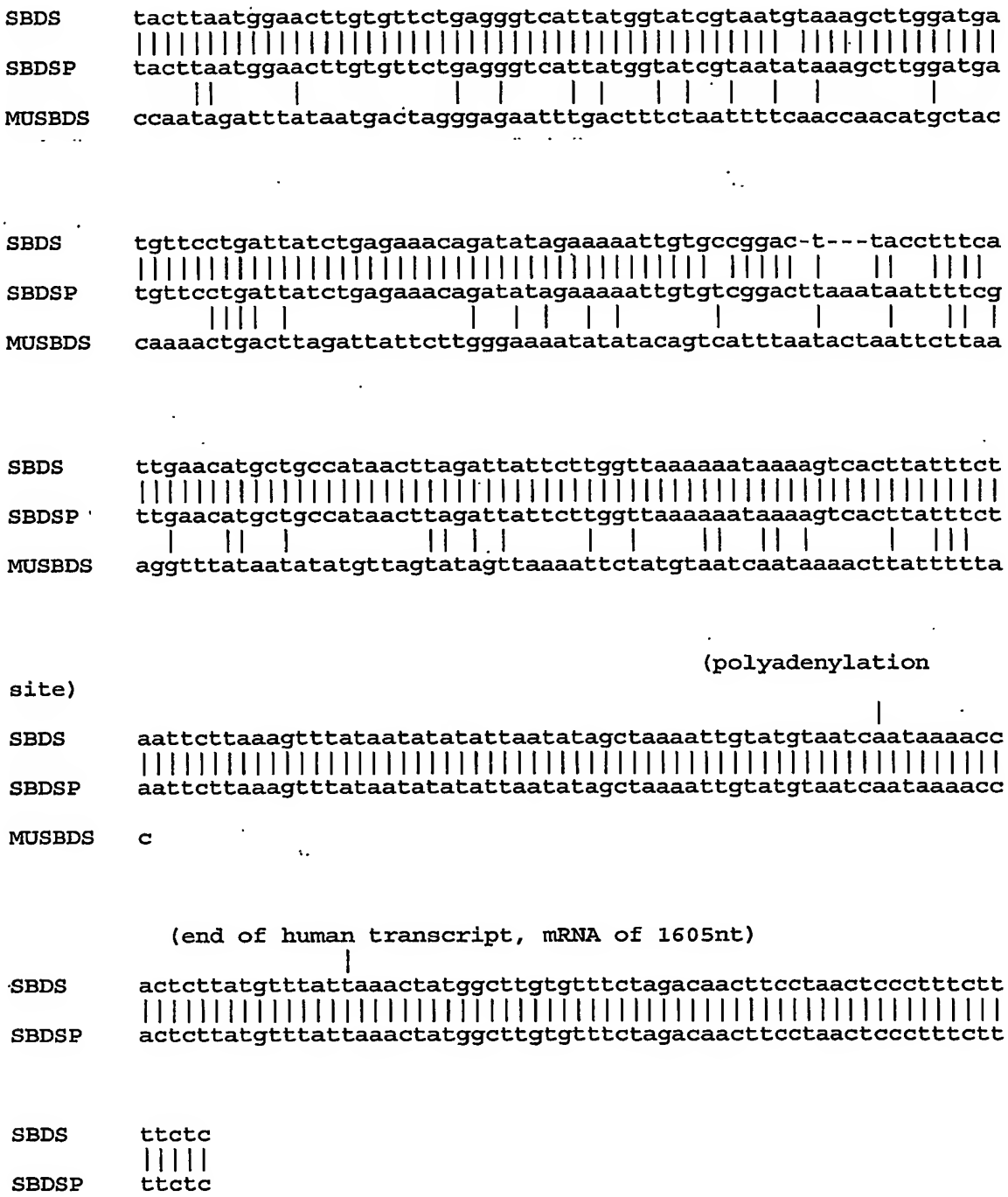
SBDS Exon 5:

```
                 Primer K (SDCR9x5CF) →
SBDS            tccactgtagatgtgaactaactcatctgacactacttgaagttctaaaatctttgcaaa
                |||||||||||||||||||||| ||||||||||||||||||||||||||||||||||||
SBDSP           tccactgtagatgtgaactaacccatctgacactacttgaagttctaaaatctttgcaaa
                          |         |  |         |    | | |   |   |    | |
MUSBDS          gtatactgtggctgtcttcagacacagcagaaggcatcggatcccattacagatggttgt


SBDS            actgtacacatgggccaggcacagtggctcgtgcctgtaatcccagcactttgggaggcc
                ||||||||| |||||||||||||||||| | ||||||||||||||||||||||||||||
SBDSP           actgtacacgtgggccaggcacagtggctcatacctgtaatcccagcactttgggaggcc
                     ||  ||||      |   | ||      |    |                |
MUSBDS          gagccacttgtggttgctgggaattgagctcagaacctctggaagagcagccagtgctga


SBDS            aaggtgagcagataacatggtgaaaccctatctctactaaaaatacaaaaaataagccag
                 ||| |||||||||||| |||||||||||| ||||||||||||||||||||||||||||
```


SBDSP gaggcgagcagataacacggtgaaaccctgtctctactaaaaatacaaaaaataagccag
MUSBDS gcatctctacagcctctgaacccagggtcttgatgctaagcagtgctcactctcagtatg
SBDS gtgtggtggtggg-ttcctgtaatcccagtttcttgggaggctgaggcaggagaatcact
SBDSP gtgtggtggtgggcgt-ctgtaatcccagtgtcttgggaggccgaggcaggagaatcact
MUSBDS agctgcagcactggccaggtgagtcttcaagggtgtcttaatcaggcttttactgctgtg
SBDS tgaacctgggaggcggaggctgcagtgagccaagatcacaccactgcactctatctc-aa
SBDSP. tgaacctgggaggtggaggctgcagtgagccaagatcacaccactgcactctatctcaaa
MUSBDS aacagacaccaggaccaatgcaagtcttataaagaacaacatttagttgagtctggctta
SBDS aaaaaaat--aa-attaacatacacatggtgtctacataagtcttcacattgctttttct
SBDSP aaaaaaataaaacaaaaacatacacatggtgtctacgtaagtcttcacattgctttttct
MUSBDS caggttcagaggttcagtccattatcaaggtgggagcatggtagtatccaggtgggaatg
SBDS ccttcatacgtggaggtgactttactgagctataaaatgtaatgctaaattttagtatga
SBDSP ccttcatacgtggaggtgactttactgagctataaaatgtaatgctaaattttagtatga
MUSBDS atacaggaggggctgagagttcgacatcttcatctgaaggctgctagcagaatactgact
SBDS gaagaatcagagttttctagtttgtcccttccatttacagctgaagaatcagaataagtg
SBDSP gaagaatcagagttttctagtttgtcccttccatttacagcggaagaatcagaataagtg
MUSBDS. tcgaggctgttaggatgagggtcttaaagcctatgaccacagggacacaccttctaatag
SBDS tttaaacatagggattaatgccttgtcacaggggctacatggacacttgagggcagagg
SBDSP tttaaacatagggattaatgccttgtcacaggggctacatggatacttgagggcagagg
MUSBDS. tgtcactccccgggctgagcatatacaaaccgtaacacgggataagtgcctttcccaaag


SBDS ctaaactggaacccagtgtgccgccctacccattgtcttatctattgcaccatagaactg
SBDSP ctgaactggaacccagtgtgccgccctacccattgtcttatctattgcaccatagaactg
MUSBDS tccaacagtaggtgcttagaatcgagacagaaccccaggcccagcctgctgccctggcct
SDCR9x5Fseq →
SBDS tggtattattagagatctggacagcattgtgcttgcctcaaaggaagttaaagctgagtt
SBDSP tggtatta---gagatctggacagcattgtgcttgcctcaaag----ttaaagctgagtt
MUSBDS ccatgtgagcagcacctagaacacagtcatagatctgccctgagcattcaaactgggctt
625
V C
SBDS tattctgtgtcttgctcatcctcatgtggtaatctgctacgttaaatgtttcagGTATGT
SBDSP tattctgtgtcttgctcatcctcatttggtaaactgctacgttaaatgtttcaggtatgt
MUSBDS attctgtgccgatgcccatcttcccttggaaaccagctgtgttactcattgcagGTGTGC
V C
L I D P G C F R E I D E L I K K E T K G
SBDS CTGATTGACCCGGGCTGCTTCCGAGAAATTGATGAGCTAATAAAAAAGGAAACTAAAGGC
SBDSP ctgattgacctgggctgcttccgagaaattgatgagctaataaaaaaggaaaccaaaggc
MUSBDS CTCATCGACCCAGGCTGCTTCAGAGAAATTGATGAGCTAATAAAAAAGGAAACGAAAGGC
L I D P G C F R E I D E L I K K E T K G
750
K G S L E V L N L K D V E E G D E K F E
SBDS AAAGGTTCTTTGGAAGTACTCAATCTGAAAGATGTAGAAGAAGGAGATGAGAAATTTGAA
SBDSP aaaggttctttggaagtactcaatctgaaagattt-gaagaaggagatgagaaatttgaa
MUSBDS AGGGGTTCTCTGGAAGTGCTCAGTCTGAAGGACGTGGAGGAAGGCGATGAGAAGTTTGAA
R G S L E V L S L K D V E E G D E K F E
SBDS tgacacccatcaatctcttcacctctaaaacactaaagtgtttccgtttccgacggcact
SBDSP tgacacccatcagtctcttcacctctaaaacactaaagtgttttcgtttccaacagcact

```
MUSBDS   TGAcaccgcccggctcctcaactggagcacgaccgaggacgcttgttcctcacagcagca

SBDS     gtttcatgtctgtggtctgccaaatacttgcttaaactatttgacattttctatctttgt
SBDSP    gtttcatgtctgtggtctgccaaatacttgctcaaactatttgacattttctatctttgt
MUSBDS   gctcgttctgtgacctgccaaacgccctgctcacgcgacgtgccactttccatcttgtgt

SBDS     gttaacagtggacacagcaaggctttcctacataagtataataatgtgggaatgatttgg
SBDSP    gttaacagtggacacagcaaggctttcctacataagtataataatgtgggaatgatttgg
MUSBDS   taaacatttacccaggtacctgggtatttttgttgtcaattggggtttccagcaaaaatg

SBDS     ttttaattataaactggggtctaaatcctaaagcaaaattgaaactccaagatgcaaagt
SBDSP    ttttaattataaactggggtctaaatcctaaagcaaaattgaaactccaggatgcaaaat
MUSBDS   aaaaataacctaaaatacagagtccagaacagctgctcactgctgcgtctgcctttctag

         ← Primers L/R (RTSDCR95R/SDCR9x5BR)
SBDS     ccagagtggcattttgctactctgtctcatgccttgatagctttccaaaatgaaagttac
SBDSP    ccagagtggcattttgctactctgtctcatgccttgatagctttccaaaatgaaagttac
MUSBDS   ttccaggggaccagagacagcattggtggataagaaggtagagttagtccatgacagatc

SBDS     ttgaggcagctcttgtgggtgaaaagttatttgtacagtagagtaagattattaggggta
SBDSP    ttgaggcagctcttgtgggtgaaaagttttttgtacagtagagtaagattattaggggta
MUSBDS   attggagaggggtctgaataacaaaggggggtacgcctgctggaaagaagatggggtgttt

SBDS     tgtctatacaacaaaaggggggtctttcctaaaaaagaaaacatatgatgcttcatttc
SBDSP    tgtctatacgacaaaa-gggggtctttcctaaaaaagaaaac--atgatgcttcatttc
MUSBDS   ctgaataatgaagtgcaggtatggggtgtgagcatggagagaagagttcctgggtccctc
```


SBDS tacttaatggaacttgtgttctgagggtcattatggtatcgtaatgtaaagcttggatga
SBDSP tacttaatggaacttgtgttctgagggtcattatggtatcgtaatataaagcttggatga
MUSBDS ccaatagatttataatgactagggagaatttgactttctaattttcaaccaacatgctac
SBDS tgttcctgattatctgagaaacagatatagaaaaattgtgccggac-t---tacctttca
SBDSP tgttcctgattatctgagaaacagatatagaaaaattgtgtcggacttaaataatttcg
MUSBDS caaaactgacttagattattcttgggaaaatatatacagtcatttaatactaattcttaa
SBDS ttgaacatgctgccataacttagattattcttggttaaaaaataaaagtcacttatttct
SBDSP ttgaacatgctgccataacttagattattcttggttaaaaaataaaagtcacttatttct
MUSBDS aggtttataatatatgttagtatagttaaaattctatgtaatcaataaaacttatttta
(polyadenylation
site)
SBDS aattcttaaagtttataatatatattaatatagctaaaattgtatgtaatcaataaaacc
SBDSP aattcttaaagtttataatatatattaatatagctaaaattgtatgtaatcaataaaacc
MUSBDS c
(end of human transcript, mRNA of 1605nt)
SBDS actcttatgtttattaaactatggcttgtgtttctagacaacttcctaactccctttctt
SBDSP actcttatgtttattaaactatggcttgtgtttctagacaacttcctaactccctttctt
SBDS ttctc
SBDSP ttctc

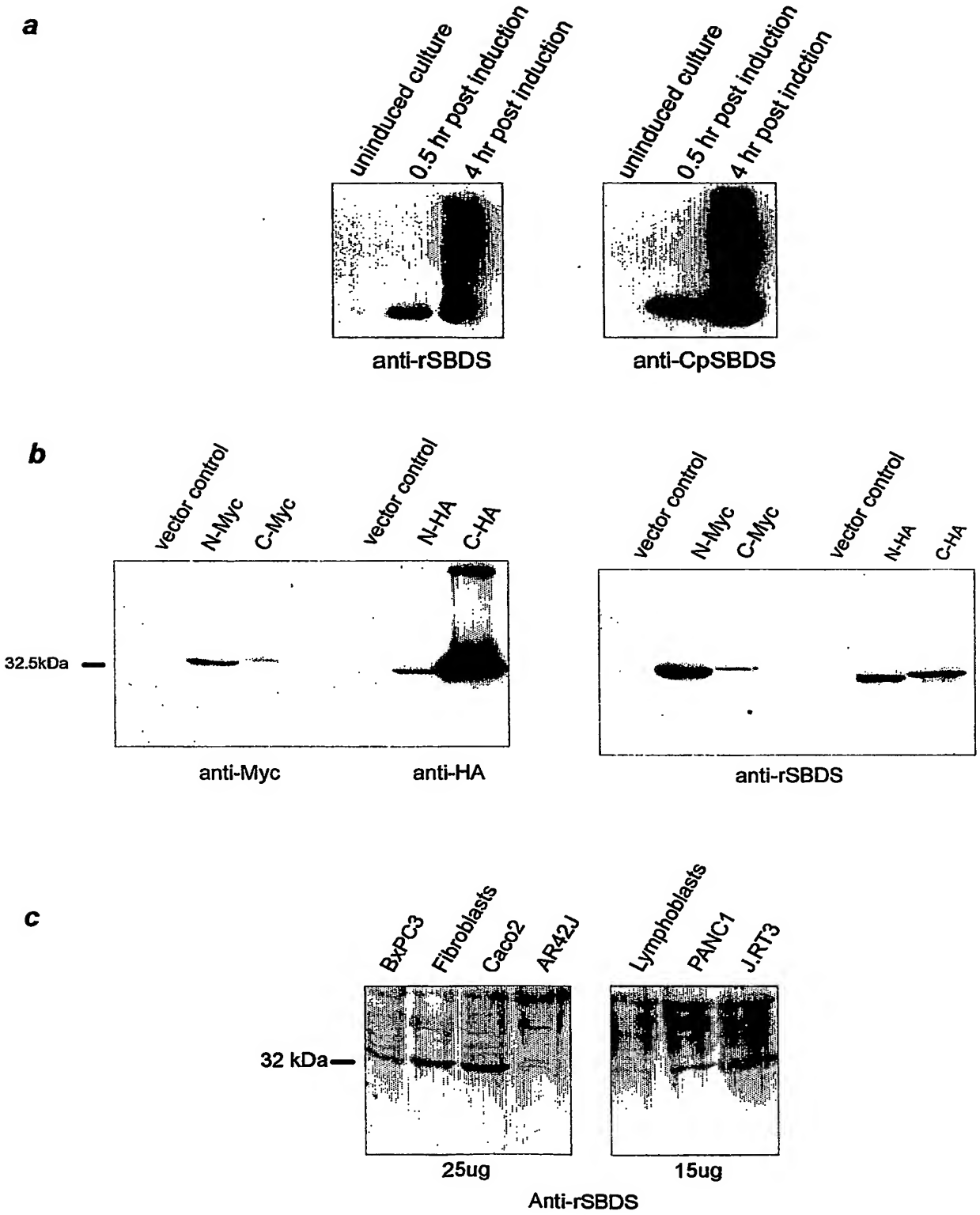

a
uninduced culture
0.5 hr post induction
4 hr post induction
anti-rSBDS
uninduced culture
0.5 hr post induction
4 hr post indction
anti-CpSBDS
b
vector control
N-Myc
C-Myc
vector control
N-HA
C-HA
32.5kDa
anti-Myc
anti-HA
vector control
N-Myc
C-Myc
vector control
N-HA
C-HA
anti-rSBDS
c
BxPC3
Fibroblasts
Caco2
AR42J
Lymphoblasts
PANC1
J.RT3
32 kDa
25ug
15ug
Anti-rSBDS

FIGURE 7